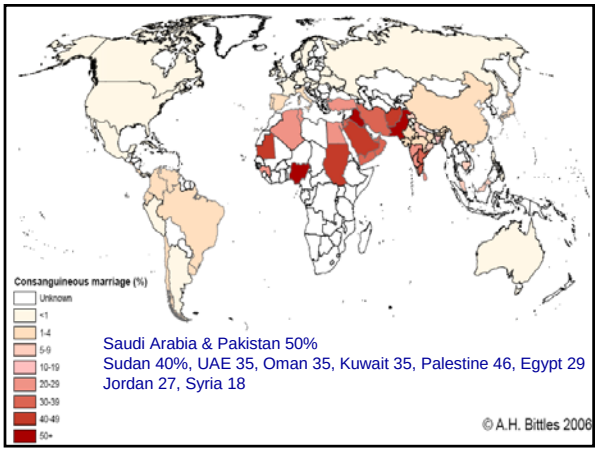
Newborn Screening In Saudi Arabia Country Report

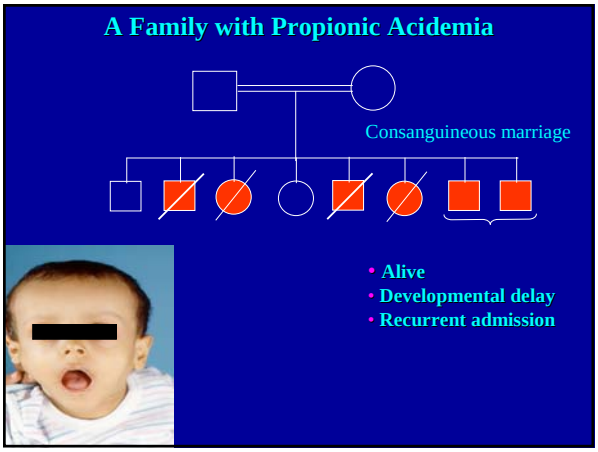
NLNBS

Mohamed S. Rashed, BPharm, MSc, PhD
Director, National Laboratory for Newborn Screening (NLNBS)
King Faisal Specialist Hospital and Research Centre

Population: 22,000,000 (16 M Saudi, 6M expatriates)
450,000 birth per year
Large family size: between 6-12



Saudi Arabia & Pakistan 50%
Sudan 40%, UAE 35, Oman 35, Kuwait 35, Palestine 46, Egypt 29
Jordan 27, Syria 18

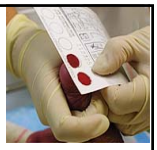


Screening Milestones for Saudi Arabia

- 1991 Cord blood Hypothyroidism screening at 20 centers
- 1994 Introduction of tandem mass spectrometry at KFSH&RC
- 1995-1998 First MS/MS pilot study, published 1999
- 1995-2005 Expanded newborn screening on small scale
- Aug 2005 National newborn screening started
- Phase I includes 24 birth centers and 120,000 newborns
- 2008 Coverage of >400,00 newborns

HRH Prince Sultan Bin Salman with Excellency Minister of Health (2004)

SNBS Phase I



- 120,000 newborn blood samples collected in first year
- 24 hospitals are included from different regions
- Screen for 16 diseases (Panel A & Panel B)
- Study logistical and coordination problems
- Outcome will lead to national registry (still pending)
- Coordinate recall and tracking problems (still a big problem)

Diseases Covered by the Saudi Newborn Screening

Panel A: Diseases Covered by Tandem Mass Spectrometry:

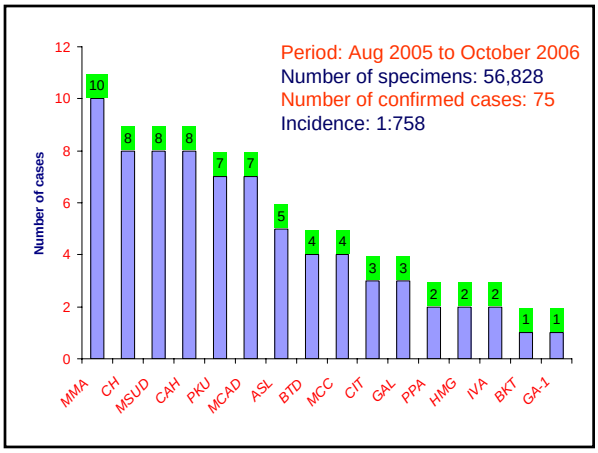
1. *Phenylketonuria (PKU)*
2. *Maple Syrup Urine Disease (MSUD)*
3. *Argininosuccinase Deficiency (ASL)*
4. *Citrullinemia (ASD)*
5. *HMG-CoA Lyase Deficiency (HMG)*
6. *Isovaleric Acidemia (IVA)*
7. *Methylmalonic Acidemia (MMA)*
8. *Propionic Acidemia (PA)*
9. *Beta-keto Thiolase Deficiency (BKT)*
10. *Glutaric Acidemia type-I (GA-I)*
11. *Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)*
12. *3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)*

Panel B: Diseases Covered by Kit-based Assays:

1. *Galactosemia*
2. *Congenital Hypothyroidism (CH)*
3. *Congenital Adrenal Hyperplasia (CAH)*
4. *Biotinidase Deficiency (BD)*

Technology

- 4 Tandem mass spectrometry (MS/MS & LC-MS/MS).
- 2 Gas chromatography mass spectrometry (GC-MS)
- 4 High pressure liquid chromatography (HPLC)
- 1 UPLC system
- 1 Autodelfia TSH & CAH
- 1 VictorD2 BTD & GALAC
- 3 Blood spots punchers
- 1 Lab Database since 1994
- Relatively small lab space



Recall is a nightmare

3 months Sample

Questionable MS/MS	Recalled	Failed Recall
90	10	80
	11%	89%
Questionable 17-OHP	**Recalled**	**Failed Recall**
217	28	189
	13%	*87%*
Questionable TSH	**Recalled**	**Failed Recall**
167	7	160
	4%	*96%*
Questionable BTD	**Recalled**	**Failed Recall**
54	2	52
	4%	*96%*
Questionable Galt	**Recalled**	**Failed Recall**
40	1	39
	2.5%	*97.5%*

Barriers to Improving SNBS

- *Incomplete or incorrect demographics*
- *Early specimen collection*
- *Communications and resource difficulties*
- *Recall difficulties*
- Inadequate oversight & tracking & medications
- Deficiency in metabolists, dietitians, genetic counselors
- Uncertainty about financing & politics
- A significant single improvement “Mandating NBS by Law”

Thank you